



## COQ6 gene

coenzyme Q6, monooxygenase

### Normal Function

The COQ6 gene provides instructions for making an enzyme that carries out one step in the production of a molecule called coenzyme Q10. Coenzyme Q10 has several critical functions in cells throughout the body. In cell structures called mitochondria, coenzyme Q10 plays an essential role in a process called oxidative phosphorylation, which converts the energy from food into a form cells can use. Coenzyme Q10 is also involved in producing pyrimidines, which are building blocks of DNA, its chemical cousin RNA, and molecules such as ATP and GTP that serve as energy sources in the cell. In cell membranes, coenzyme Q10 acts as an antioxidant, protecting cells from damage caused by unstable oxygen-containing molecules (free radicals), which are byproducts of energy production.

### Health Conditions Related to Genetic Changes

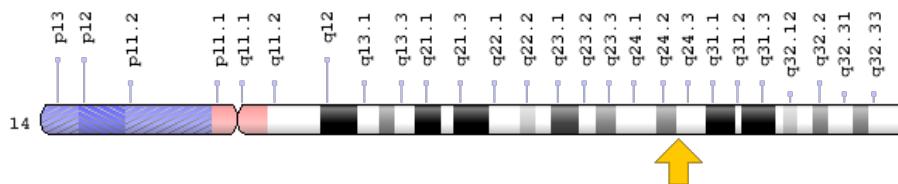
#### primary coenzyme Q10 deficiency

At least seven mutations in the COQ6 gene have been found to cause a disorder known as primary coenzyme Q10 deficiency. This rare disease usually becomes apparent in infancy or early childhood, but it can occur at any age. It can affect many parts of the body, most often the brain, muscles, and kidneys. The COQ6 gene mutations associated with this disorder result in a COQ6 enzyme with an abnormal structure that likely impairs its function, leading to reduced production of coenzyme Q10. Studies suggest that a shortage (deficiency) of coenzyme Q10 impairs oxidative phosphorylation and increases the vulnerability of cells to damage from free radicals. A deficiency of coenzyme Q10 may also disrupt the production of pyrimidines. These changes can cause cells throughout the body to malfunction, which may help explain the variety of organs and tissues that can be affected by primary coenzyme Q10 deficiency.

## Chromosomal Location

Cytogenetic Location: 14q24.3, which is the long (q) arm of chromosome 14 at position 24.3

Molecular Location: base pairs 73,949,934 to 73,963,112 on chromosome 14 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CGI-10
- CGI10
- coenzyme Q6 homolog, monooxygenase
- coenzyme Q10 monooxygenase 6
- COQ10D6
- ubiquinone biosynthesis monooxygenase COQ6, mitochondrial isoform a
- ubiquinone biosynthesis monooxygenase COQ6, mitochondrial isoform b

## Additional Information & Resources

### Educational Resources

- Linus Pauling Institute: Coenzyme Q10  
<http://lpi.oregonstate.edu/mic/dietary-factors/coenzyme-Q10>
- Molecular Biology of the Cell (fourth edition, 2002): How Cells Obtain Energy From Food  
<https://www.ncbi.nlm.nih.gov/books/NBK26882/>
- The Cell: A Molecular Approach (second edition, 2000): The Mechanism of Oxidative Phosphorylation  
<https://www.ncbi.nlm.nih.gov/books/NBK9885/>

## GeneReviews

- Primary Coenzyme Q10 Deficiency  
<https://www.ncbi.nlm.nih.gov/books/NBK410087>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28COQ6%5BTIAB%5D%29+OR+%28coenzyme+Q6,+monooxygenase%5BTIAB%5D%29%29+OR+%28coenzyme+Q10+monooxygenase+6%5BTIAB%5D%29+OR+%28coenzyme+Q6+homolog,+monooxygenase%5BTIAB%5D%29+OR+%28ubiquinone+biosynthesis+monooxygenase+COQ6,+mitochondrial+isoform+a%5BTIAB%5D%29+OR+%28ubiquinone+biosynthesis+monooxygenase+COQ6,+mitochondrial+isoform+b%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

## OMIM

- COQ6, S. CEREVISIAE, HOMOLOG OF  
<http://omim.org/entry/614647>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=COQ6%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=20233](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=20233)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/51004>
- UniProt  
<http://www.uniprot.org/uniprot/Q9Y2Z9>

## **Sources for This Summary**

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*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4112527/>
- GeneReview: Primary Coenzyme Q10 Deficiency  
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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21540551>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3083770/>

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